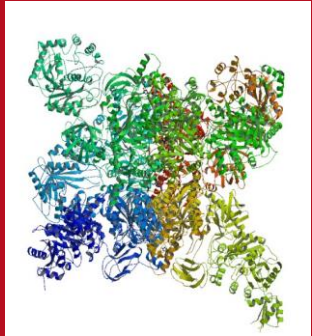




THE OHIO STATE UNIVERSITY



# Propionic Acidemia

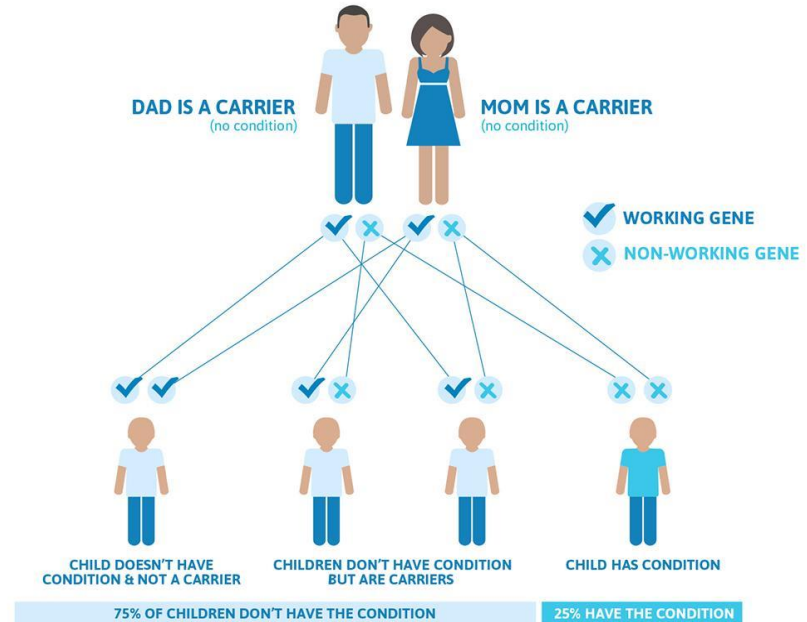
Nathan Kudlapur, Rosalyn Liou, Grant Barnett,  
Josh Shekhtman, Jake Dinkins, Jeff Lotthammer,  
Kris Skugor, Pho Lee, Tommy Bodnar

*Service Learning Initiative in Biochemistry 5614 (Autumn 2018)*

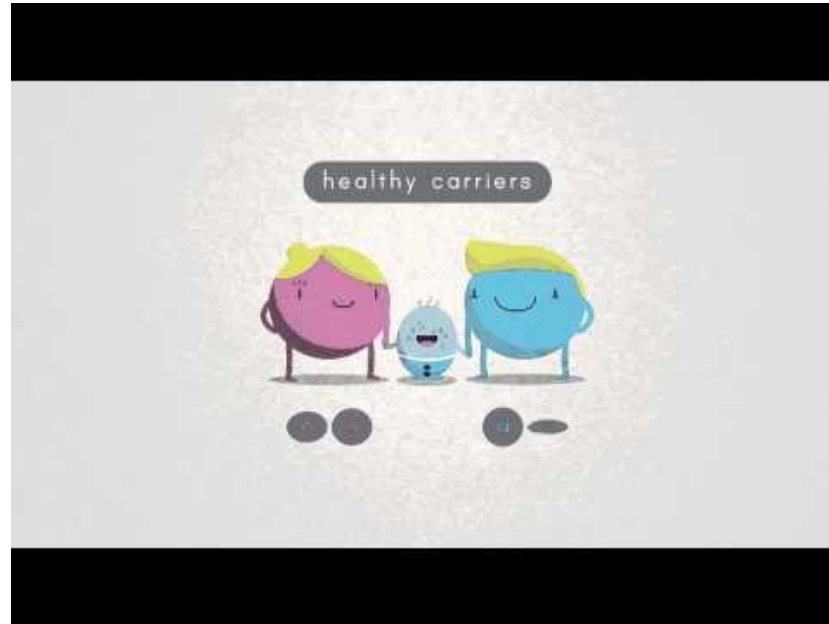
# History, Prevalence, and Genetic Basis

- First report in 1961 - deficiency of propionyl-CoA carboxylase
- Prevalence : 1/50,000 live births (US)
- Higher prevalence in Japan and some Amish communities (1/17,000)
- Autosomal recessive disorder

## Autosomal Recessive Inheritance Pattern

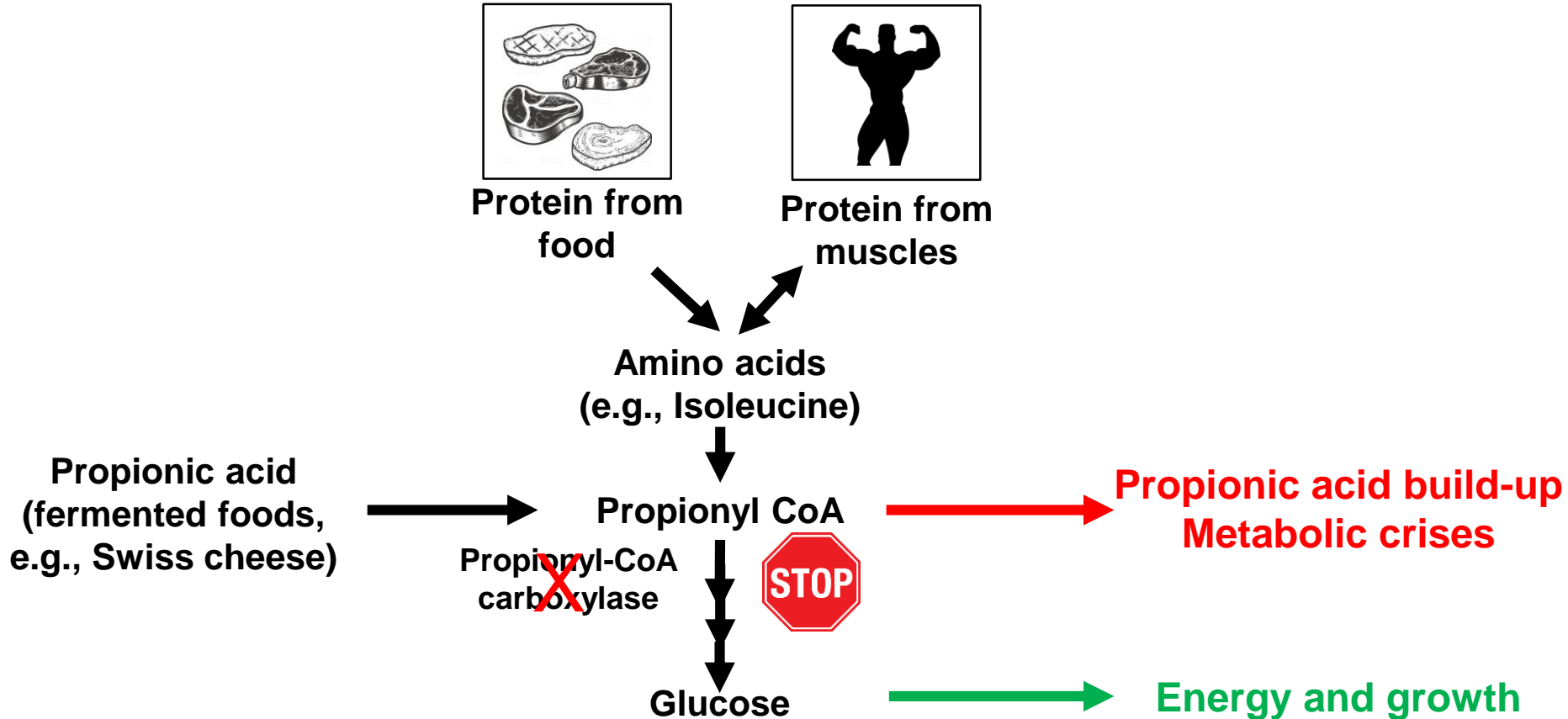


# Autosomal Recessive Inheritance



HNEkidshealth. "Autosomal Recessive Inheritance - Genetics." YouTube, YouTube, 30 Mar. 2015, [www.youtube.com/watch?v=Nv6qUsKYodA](http://www.youtube.com/watch?v=Nv6qUsKYodA).

# Biochemical Features



# Symptoms

- **Type I: Early onset (within a few days of birth)**

- Poor feeding
- Vomiting
- Weak muscle tone
- Dehydration



Reflux or Just a Sick Baby? Digital image retrieved Oct. 30, 2018 from <https://madabouttheboys.net/reflux-or-just-a-sick-baby/>

- **Type II: Late onset (childhood)**

- Similar symptoms to those of early onset
- Intellectual disability
- Delayed development

# Neonatal Diagnosis

## Neonatal Diagnosis (after birth)

- Mass spectrometry
  - Measure metabolites indicative of the disease (propionic acid build-up)
- Additional lab test
  - Measure amount of enzymatic activity in leukocytes or cultured fibroblasts



Texas Newborn Screening - Info for Parents. Digital image retrieved Oct. 30, 2018 from <https://www.dshs.texas.gov/newborn/expandparent.shtm>



The Johns Hopkins University. Digital image retrieved Oct. 30, 2018 from [https://www.hopkinsmedicine.org/news/media/releases/experts\\_recommend\\_fewer\\_lab\\_tests\\_for\\_hospitalized\\_patients](https://www.hopkinsmedicine.org/news/media/releases/experts_recommend_fewer_lab_tests_for_hospitalized_patients)

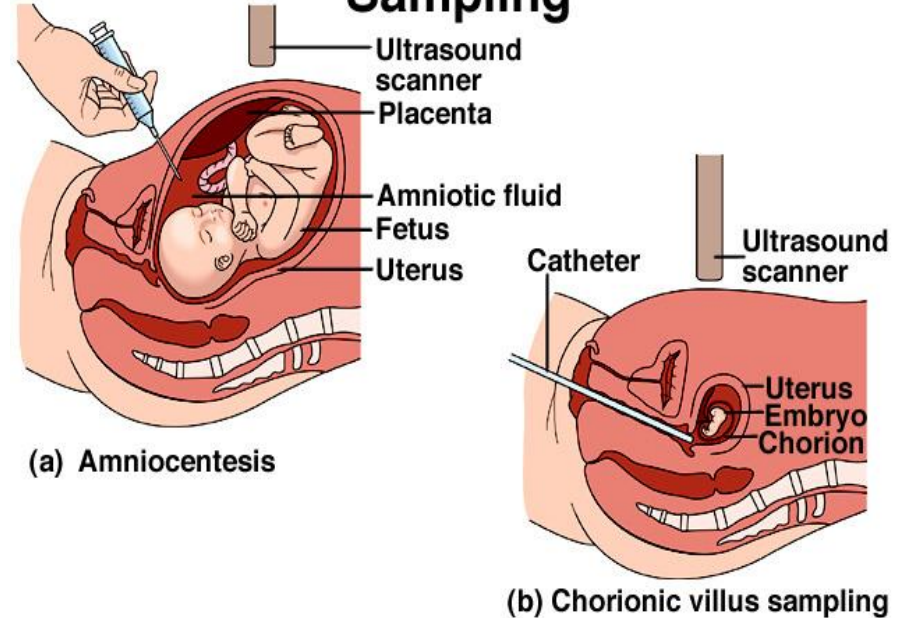
# Prenatal Diagnosis

## Prenatal diagnosis (before birth)

- Measure the concentration of propionic acid in amniotic fluid or the activity of the PCC enzyme
  - Amniocentesis
  - Chorionic Villus Sampling

Byer/Shainberg/Galliano *Dimensions Of Human Sexuality*, 5e. Copyright © 1999. The McGraw-Hill Companies, Inc. All Rights Reserved.

## Amniocentesis & Chorionic Villus Sampling



Digital image retrieved Oct. 30, 2018.

# Prognosis

- **Infants treated early can develop and grow normally**
  - This is why propionic acidemia is part of the newborn screening panel
  - The success of treatment largely depends on detecting the disorder before a metabolic crisis
- **If a metabolic crisis does occur, effects can be much more serious:**
  - Seizures and cognitive deficiencies
  - Brain damage (especially severe cases)



# Therapy

- **For mild cases:**
  - Strict low-protein diet
    - Formula deficient in branched chain amino acids but supplemented by natural protein in predetermined amounts
  - Antibiotics
    - Suppress gut bacteria and decrease propionic acid levels
  - Avoidance of long periods of fasting

**Note:** Mild cases are generally a result of early and consistent management



Digital image retrieved Oct. 30, 2018 from <https://abspacecakes.com/blogs/protein-pancakes-recipes/do-low-carb-diets-work>

Digital image retrieved Oct. 30, 2018 from <https://medium.com/@thryve/the-development-of-the-gut-microbiome-f060a6ba41bf>

# Therapy

- **For severe cases:**
  - Liver transplantation
    - In cases of frequent/severe metabolic decompensation despite good therapy
  - Dialysis

**Note:** Severe cases can be the result of the chronic progressive form (from a certain mutation)

# Support



- The Organic Acidemia Association is a one-stop resource for the families of those diagnosed with organic acidemia disorders.
- The Propionic Acidemia Foundation is another excellent resource at [www.pafoundation.com](http://www.pafoundation.com)
- Available services include:
  - Information (Brochures, tri-annual newsletter, recipes/meal planning)
  - Connection (Online forum, parent matching)
  - Conferences
  - Advocacy

<https://www.oaanews.org/>

# Propionic Acidemia: Project Team

Student	Project Role
Nathan Kudlapur (Text)	Genetic Basis
Rosalyn Liou	History
Pho Lee	History
Josh Shekhtman (Illustrations)	Biochemical Features
Jake Dinkins	Symptoms
Jeff Lotthammer	Diagnosis
Grant Barnett	Prognosis
Kris Skugor	Therapy
Tommy Bodnar	Support